

We claim:

1. An isolated DNA sequence up to 20 kb in length comprising a nucleic acid sequence as shown in SEQ ID NO. 1.

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2. The isolated nucleic acid of claim 1, wherein said nucleic acid is cDNA.

3. A method for diagnosing a patient as having an increased risk of developing HH disease, comprising:

providing DNA or RNA from the individual; and

assessing the DNA or RNA for the presence or absence of a base mutation at position 734 (A734C) of the SLC11A3 gene wherein the absence of the base mutation indicates the absence of a HH gene mutation in the genome of the individual and the presence of the base mutation indicates the presence of the HH gene mutation and an increase risk of developing HH disease in the genome of the individual being diagnosed.

20 4. The method of claim 3, wherein the assessing step is performed by a process which comprises subjecting the DNA or RNA to amplification using oligonucleotide primers flanking the base-pair mutation.

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5. The method of claim 4, wherein the assessing step further comprises an oligonucleotide ligation assay.

6. An oligonucleotide of at least 8 consecutive nucleotides
5 selected from a sequence unique to SEQ ID NO. 1 or the complement
of SEQ ID NO. 1.

7. The oligonucleotide of claim 6, wherein the oligonucleotide
is a member of an oligonucleotide pair for amplification of an HH
10 nucleic acid sequence.

8. An isolated polypeptide comprising the amino acid sequence
as shown in SEQ ID NO. 2.

15 9. An isolated polypeptide comprising the amino acid
sequence as shown in SEQ ID NO. 3.

20 10. The isolated polypeptide of claim 9, in which the amino
acid residue of position 734 (A734C) in exon 5 is changed from
asparagine to histidine.

11. The isolated polypeptide of claim 10, which interacts with
Hephaestin.

12. The isolated polypeptide of claim 9, which is membrane-associated.

5 13. The isolated polypeptide of claim 9, which is fused with a heterologous polypeptide.

14. The isolated polypeptide of claim 9, which is a naturally occurring polypeptide.

10 15. The isolated polypeptide of claim 9, which is produced by a recombinant DNA method.

15 16. The isolated polypeptide of claim 9, which is produced by a chemical synthetic method.

17 17. A pharmaceutical composition comprising the isolated polypeptide of claim 9.

20 18. An isolated polypeptide comprising the amino acid sequence as shown in SEQ ID NO. 4.

19. The isolated polypeptide of claim 18, in which the amino acid residue of position 734 (A734C) in exon 5 is any amino acid

except asparagine.

20. The isolated polypeptide of claim 19, which interacts with Hephaestin.

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21. The isolated polypeptide of claim 18, which is membrane-associated.

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22. The isolated polypeptide of claim 18, which is fused with a heterologous polypeptide.

23. The isolated polypeptide of claim 18, which is a naturally occurring polypeptide.

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24. The isolated polypeptide of claim 18, which is produced by a recombinant DNA method.

25. The isolated polypeptide of claim 18, which is produced by a chemical synthetic method.

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26. A pharmaceutical composition comprising the isolated polypeptide of claim 18.

27. A kit for the detection of the presence or absence of a

base mutation at position 734 (A734C) of the SLC11A3 gene.

28. The kit of claim 27, further comprising primers for amplifying the DNA containing the base-pair polymorphism at 5 position 734 (A734C) of the SLC11A3 gene.

29. A genetic marker predictive of a hereditary hemochromatosis (HH) gene mutation comprising a partial sequence of SEQ ID NO. 1 and sequences complementary therewith.

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